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| Library concerned: Brighton and Sussex NHS Library and Knowledge Service  Name of case study: Diagnosing dystroglycanopathy  Date of interview: | |
| **Interviewee** | Name:  Job Title: Consultant in Neonatology  Trust/Employing body: Brighton and Sussex University Hospitals NHS Trust  Email:  Tel: |
| **Please note: you should only provide interviewee details here if consent**  **to share on the national blog has been obtained** |
| **S****ummary of problem or reason for enquiry** | I was treating a newborn baby who I suspected may be suffering from dystroglycanopathy, a rare form of congenital muscular dystrophy that is difficult to diagnose. I needed evidence for how best to establish a firm diagnosis. |
| **Brief description of the information found / service provided** | Our clinical librarian is embedded into the neonatology service, regularly attending meetings where challenging patients are discussed, and was therefore on hand to track down the research rapidly. He carried out an evidence search and found a range of relevant institutional publications and original research. |

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| **Summary of outcome and impact** | The information you provided on genetic test options gave me what I needed to accurately diagnose this newborn patient. |
| **Immediate Impact**  **(including quotes)** | “The review articles you provided contained genetic test options and guided us on how to proceed. They helped me to explain the situation to the parents and make a plan for finding a diagnosis.” |
| **Probable future Impact**  **(including quotes)** |  |